Rare Bone Marrow Disease With Only One Cure

Family Uses Genetic Engineering to Save Daughter's Life

By FARNAZ JAVID

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Within hours of Katie Trebing's birth Dec. 12, 2002, she needed a blood transfusion to save her life -- the first of many to come. Steve and Stacy Trebing's daughter was born with Diamond Blackfan anemia, a rare bone marrow disease that affects just 30 out of every 4 million newborns each year in the United States and Canada.

Stacy has not forgotten the day her pediatrician broke the news.

"I remember vividly being in his office, holding Katie, and him saying, 'You're gonna be tied to hospitals for the rest of your life.'"

Katie's body would never make any red blood cells to carry oxygen to her organs. The Trebings' took Katie to see Diamond Blackfan anemia specialist Dr. Jeffrey Lipton, who by chance practices less than a half hour from their Long Island, N.Y., home. Stacy taped the conversation because there was so much information to absorb.

"He spent five hours with us explaining Diamond Blackfan anemia, explaining the prevalence, explaining what it does to your body, explaining the possible treatment choices, explaining what each treatment choice would, could or may or would do to Katie," she said.

Lipton told the Trebings that Katie needed transfusions every three to four weeks or she would die.

"It is a lifelong process," said . Lipton. "It can be devastating for the patient."

The transfusions would cause iron to build up in her organs, starting with the liver and heart. The damage meant a shorter life □ more than 40 percent of Diamond Blackfan patients using transfusion therapy die by the age of 40.

Oral steroids were a second treatment option that could spur Katie's body into producing red blood cells. But the drugs would cause weight gain and could stunt growth, and came with a laundry list of major long-term side effects.

Only One Cure

There was one way to cure Katie and allow her to live a normal life: a bone marrow transplant from a perfectly matched sibling. But Katie's older brother Calvin was not a match. They would need to give Katie another sibling. Lipton told them about a specific process that would ensure that their next child, and Katie, would share the same bone marrow DNA. To Steve, the whole thing seemed more like science fiction.

"It was surreal. I mean, I thought it was just in the movies," he said.

There were several steps, starting with in vitro fertilization to help the Trebings produce a maximum number of embryos. Then a doctor would test for embryos with matching bone marrow DNA using a procedure called preimplantation genetic diagnosis, or PGD. The test is done by pulling one cell from each embryo and identifying the ones that match Katie's bone marrow DNA.

"It seemed like a win-win situation," said Stacy.

She and Steve had always hoped to have a third child. They decided to move forward with the IVF using PGD. At the same time, the Trebings decided to go public. Newsday reporter Beth Whitehouse chronicled their story for the next three years.

"They allowed us to see everything -- every & bad moment, every good moment. They never made us leave," Whithouse said. As they began the IVF process, the Trebings got more bad news. Katie's latest tests showed iron from the transfusions built up in her liver more rapidly than expected. Her doctor added a new medication to help her body discharge the excess iron. The drug was pumped through her system through a needle her parents injected in her thigh. The traumatic and painful treatment lasted 10 hours a night, five nights a week.

"She despised that," said Steve.

But during all this, there was also some remarkable news -- Stacy learned she was pregnant.

On May 4, 2005, Christopher Trebing was born.

"He was always smiling, never cranky & the perfect baby," remembered Stacy.

She and Steve hoped the perfect baby would lead to the perfect cure for Katie. But they battled with the decision of whether to go forward with Katie's bone marrow transplant. Although they knew it could cure her, it could also kill her.

A Difficult Decision

"We would lie in bed and constantly torture ourselves by saying, 'What if this and what if that?'" said Steve.

Finally they made a decision to move forward with the transplant. They also decided to have one of Katie's ovaries frozen so she might be able to have biological children in the future.

On May 15, 2006, Katie checked into Memorial Sloan-Kettering Cancer Center where she would be

confined to her room until after the transplant. Doctors gave her 10 days of chemotherapy to kill her immune system so it wouldn't reject her baby brother's bone marrow. The chemo was painful and frightening, both for Katie and the Trebings.

"To give her this poison, it was very difficult," Stacy said. "You just pray you're doing the right thing."

Then on May 25, doctors were ready to do the transplant. They removed the bone marrow from 1-year-old Christopher's hip while he was under general anesthetic. Then they transfused it into Katie and the suspenseful waiting began. She lost her hair, and grew weak and pale from the effects of the chemo. But less than two weeks later, Katie began producing red blood cells for the first time ever. Thirty-seven days after checking into the hospital, Katie went home.

"Katie was just so happy," Stacy said.

The post-transplant recovery was difficult and long. Initially Katie had to go back to the hospital for check-ups three times a week. She had to take nine medications at all times of the day and night to help her body accept the transplant. Her fragile immune system meant everything needed to be sterilized. To keeps germs from spreading, Stacy kept hand sanitizer all over the house and Katie used her own bathroom.

But finally, in July 2007, doctors took Katie off all medications and told the Trebings she no longer had Diamond Blackfan anemia. Katie is cured but still sees her doctors for check-ups. Just this month Katie celebrated her fifth birthday, her first free of the cloud of disease.

"She just loves life,"said Steve.

While the Trebings received tremendous support from most people, others have accused them of creating a donor baby. Some critics say PGD creates children for "spare parts."

But for Steve and Stacy, the technology meant a healthy sibling added to their family and the chance for Katie to live a normal life.

To learn more about Diamond Blackfan anemia, please click here